Propionic acidemia: A retrospective study of 30 pediatric patients in Tunisia

A Ben Chehida 1,2,3 - Kh Gaaloul 1 - R Mezzi 1, H Boudabous 2,1,3 - S Ben Mrad 1 - M Zribi 1 - S Ben Messaoud 1 – R Ben Abdelaziz 1 - H Azzouz 1 – MS Abdelmoula 1,3 - N Tebib 1,3,2

1: La Rabta Hospital; Tunis- Tunisia
2: Tunisian Association for the Study of Inherited Errors of Metabolism;
3: Research Laboratory LR12SP02
Propionic acidemia: A retrospective study of 30 pediatric patients in Tunisia – Ben Chehida et al, Tunisia

**AIM**

To describe the epidemiology of propionic acidemia (PA) and its outcome, as they are still unknown in Tunisia

**METHODS**

- **Type of study**: Cross sectional retrospective (medical records)

- **Inclusion criteria**:  
  - patients with PA (confirmed by OA profile in urine)
  - managed in La Rabta Hospital,

- **Time of study**: 2000 to 2020

**RESULTS:**

30 patients (40% ♀ - 60% ♂)

*Family history* (infant deaths, similar presentation): 14 cases

*Consanguinity*: 19 cases
Propionic acidemia: A retrospective study of 30 pediatric patients in Tunisia – Ben Chehida et al, Tunisia

RESULTS:
Features at presentation

Targeted neonatal screening (2 cases):

asymptomatic ++

Early onset form (70%, neonatal onset: 63%):

- Hematologic abnormalities: 58%
- Hyperammonemia: 90%
- Acidosis: 95%
- Hypoglycemia: 26%
- Vomiting: 26%
- Dehydration: 53%
- Feeding difficulties: 47%
- Ketonuria: 74%
- Respiratory distress: 68%
- Neurological distress: 90%

Late onset form (30%):

- Hematologic anomalies: 33%
- Hyperammonemia: 44%
- Ketoacidosis: 44%
- Growth impairment: 33%
- Neurological symptoms: 66%
RESULTS: Management

- Low protein (natural) diet: 100%
- Valine-free formula: 57%
- Oral metronidazole: 100%
- L-carnitine: 67%
- Biotine: 23%

RESULTS: Outcome

- Duration of follow-up = 1 year to 17 years
- Mortality rate: 50%
- Patients lost of view: 30%
- Many complications (figure 2)
Propionic acidemia: A retrospective study of 30 pediatric patients in Tunisia – Ben Chehida et al, Tunisia

RESULTS: Complications

figure 2: Complications

<table>
<thead>
<tr>
<th>Condition</th>
<th>Percentage</th>
</tr>
</thead>
<tbody>
<tr>
<td>Stunted growth</td>
<td>13%</td>
</tr>
<tr>
<td>Learning disabilities</td>
<td>30%</td>
</tr>
<tr>
<td>Abnormal EEG</td>
<td>62%</td>
</tr>
<tr>
<td>Abnormal MRI</td>
<td>40%</td>
</tr>
<tr>
<td>Abnormal neurological development</td>
<td>3%</td>
</tr>
<tr>
<td>Pancreatitis</td>
<td>42%</td>
</tr>
<tr>
<td>Decompensations</td>
<td>77%</td>
</tr>
</tbody>
</table>

RESULTS: Complications

CONCLUSION

In Tunisia, the outcome of propionic acidemia is still unfavorable.

A better awareness concerning diagnosis, management and monitoring is warranted.